chromosome 3

Humans normally have 46 chromosomes in each cell, divided into 23 pairs. Two copies of chromosome 3, one copy inherited from each parent, form one of the pairs. Chromosome 3 spans about 198 million base pairs (the building blocks of DNA) and represents approximately 6.5 percent of the total DNA in cells.

Identifying genes on each chromosome is an active area of genetic research. Because researchers use different approaches to predict the number of genes on each chromosome, the estimated number of genes varies. Chromosome 3 likely contains 1,000 to 1,100 genes that provide instructions for making proteins. These proteins perform a variety of different roles in the body.

Health Conditions Related to Chromosomal Changes

The following chromosomal conditions are associated with changes in the structure or number of copies of chromosome 3.

3p deletion syndrome

3p deletion syndrome is a condition that often results in intellectual disability, developmental delay, and abnormal physical features. 3p deletion syndrome is caused by the deletion of the end of the small (p) arm of chromosome 3. The size of the deletion varies among affected individuals from approximately 150,000 DNA building blocks (base pairs) to 11 million base pairs and can include four to 71 known genes. In some individuals, the deletion involves material near the end of the chromosome but does not include the tip (the telomere).

The signs and symptoms related to 3p deletion syndrome result from the loss of genes in the 3p region; however, it is difficult to determine which genes influence specific features because not all affected individuals are missing the same genes.

cancers

Changes in chromosome 3 have been identified in a type of kidney cancer called clear cell renal carcinoma. This cancer can develop when one copy of chromosome 3 is missing or when part of the p arm of chromosome 3 is deleted. Additionally, clear cell renal carcinoma can be associated with abnormal exchanges of genetic material, called translocations, between chromosome 3p and another chromosome. Unlike the change that causes 3p deletion syndrome (described above), the genetic changes associated with clear cell renal carcinoma are somatic, which means they are acquired during a person's lifetime and are present only in certain kidney cells.

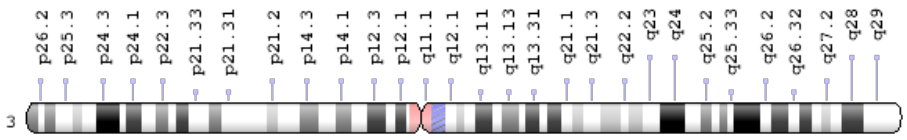
These genetic changes allow the cells to grow and divide in an uncontrolled way to form a tumor.

other chromosomal conditions

Other changes in the structure of chromosome 3 can have a variety of effects, including intellectual disability, developmental delay, distinctive facial features, birth defects, and other health problems. Changes to chromosome 3 include extra (duplicated) or deleted segments of the p arm or long (q) arm of the chromosome in each cell. As in 3p deletion syndrome (described above), the size of the extra or deleted segments varies; the amount of genetic material involved contributes to the signs and symptoms that develop. Rarely, chromosome 3 can form a circular structure called a ring chromosome, which occurs when a chromosome breaks in two places and the ends of the chromosome arms fuse together. When the ring chromosome forms, genes near the ends of chromosome 3 are deleted, and because of the ring shape, the chromosome cannot copy (replicate) itself normally during cell division, likely contributing to health problems.

Chromosome Diagram

Geneticists use diagrams called idiograms as a standard representation for chromosomes. Idiograms show a chromosome's relative size and its banding pattern, which is the characteristic pattern of dark and light bands that appears when a chromosome is stained with a chemical solution and then viewed under a microscope. These bands are used to describe the location of genes on each chromosome.



Credit: Genome Decoration Page/NCBI

Additional Information & Resources

MedlinePlus

- Encyclopedia: Chromosome
  https://medlineplus.gov/ency/article/002327.htm

Additional NIH Resources

- National Cancer Institute: Renal Cell Cancer Treatment
  https://www.cancer.gov/types/kidney/hp/kidney-treatment-pdq
- National Human Genome Research Institute: Chromosome Abnormalities
  https://www.genome.gov/11508982/
- The Cancer Genome Atlas: Clear Cell Kidney Carcinoma
  https://cancergenome.nih.gov/cancersselected/kidneyclearcell

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Chromosomes,+Human,+Pair+3%5BMAJR%5D%29+AND+%28Chromosome+3%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- RENAL CELL CARCINOMA, NONPAPILLARY
  http://omim.org/entry/144700

Sources for This Summary

- Brunelli M, Fiorentino M, Gobbo S, Sperandio N, Cheng L, Cossu-Rocca P, Segala D, Eble JN, Delahunt B, Novara G, Ficarra V, Martignoni G. Many facets of chromosome 3p cytogenetic findings in clear cell renal carcinoma: the need for agreement in assessment FISH analysis to avoid diagnostic errors. Histol Histopathol. 2011 Sep;26(9):1207-13. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21751152
- Ensembl Human Map View: Chromosome 3
  http://www.ensembl.org/Homo_sapiens/Location/Chromosome?chr=3;r=3:1-198022430
- Goobie S, Knijnenburg J, Fitzpatrick D, Sharkey FH, Lionel AC, Marshall CR, Azam T, Shago M, Chong K, Mendoza-Londono R, den Hollander NS, Ruivenkamp C, Maher E, Tanke HJ, Szuhai K, Wintle RF, Scherer SW. Molecular and clinical characterization of de novo and familial cases with microduplication 3q29: guidelines for copy number variation case reporting. Cytogenet Genome Res. 2008;123(1-4):65-78. doi: 10.1159/000184693. Epub 2009 Mar 11.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19287140
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2920184/
- Guilherme RS, Bragagnolo S, Pellegrino R, Christofolini DM, Takeno SS, Carvolheira GM, Kulikowski LD, Melaragno MI. Clinical, cytogenetic and molecular study in a case of r(3) with 3p deletion and review of the literature. Cytogenet Genome Res. 2011;134(4):325-30. doi: 10.1159/000329478. Epub 2011 Aug 17. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21849783
- Malmgren H, Sahlén S, Wide K, Lundvall M, Blennow E. Distal 3p deletion syndrome: detailed molecular cytogenetic and clinical characterization of three small distal deletions and review. Am J Med Genet A. 2007 Sep 15;143A(18):2143-9.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17696125

- Map Viewer: Genes on Sequence
  https://www.ncbi.nlm.nih.gov/mapview/maps.cgi?ORG=human&MAPS=ideogr,ugHs,genes&CHR=3
- Muzny DM, Scherer SE, Kaul R, Wang J, Yu J, Sudbrak R, Buhay CJ, Chen R, Cree A, Ding Y, Dugan-Rocha S, Gill R, Gunaratne P, Harris RA, Hawes AC, Hernandez J, Hodgson AV, Hume J, Jackson A, Khan ZM, Kovar-Smith C, Lewis LR, Lozado RJ, Metzker ML, Milosavljevic A, Miner GR, Morgan MB, Nazareth LV, Scott G, Sodergren E, Song XZ, Steffen D, Wei S, Wheeler DA, Wright MW, Worley KC, Yuan Y, Zhang Z, Adams CQ, Ansari-Lari MA, Ayele M, Brown MJ, Chen G, Chen Z, Clendenning J, Clerc-Blankenburg KP, Chen R, Chen Z, Davis C, Delgado O, Dinh HH, Dong W, Draper H, Ernst S, Fu G, Gonzalez-Garay ML, Garcia DK, Gillett W, Gu J, Hao B, Haugen E, Havlak P, He X, Hennig S, Hu S, Huang W, Jackson LR, Jacob LS, Kelly SH, Kube M, Levy R, Li Z, Liu B, Liu J, Liu W, Lu J, Maheshwari M, Nguyen BV, Okwuonu GO, Palmeiri A, Pasternak S, Perez LM, Phelps KA, Plopper FJ, Qiang B, Raymond C, Rodriguez R, Saenphimmachak C, Santibanez J, Shen H, Shen Y, Subramanian S, Tabor PE, Verduzco D, Waldron L, Wang J, Wang J, Wang Q, Williams GA, Wong GK, Yao Z, Zhang J, Zhang X, Zhao G, Zhou J, Zhou Y, Nelson D, Lehrach H, Reinhardt R, Naylor SL, Yang H, Olson M, Weinstock G, Gibbs RA. The DNA sequence, annotation and analysis of human chromosome 3. Nature. 2006 Apr 27;440(7088):1194-8.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16641997
- Pohjola P, de Leeuw N, Penttinen M, Kääriäinen H. Terminal 3p deletions in two families--correlation between molecular karyotype and phenotype. Am J Med Genet A. 2010 Feb;152A(2): 441-6. doi: 10.1002/ajmg.a.33215.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20101686
- UCSC Genome Browser: Statistics
  http://genome.cse.ucsc.edu/goldenPath/stats.html

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/chromosome/3.pdf

Reviewed: June 2016
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services